CORPUS CALLOSUM DYSGENESIS AND COLPOCEPHALY

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ABSTRACT

We present the case of T.F., a 28 year-old woman diagnosed with corpus callosum dysgenesis and colpocephaly. The diagnosis is based on cerebral imaging – magnetic resonance imaging (MRI) and computerised tomography (CT).

Key words: corpus callosum, dysgenesis, colpocephaly

INTRODUCTION

The **corpus callosum** is a band of approximately 200 million neural fibers that connects the left and right cerebral hemispheres.

It begins to develop around the 12th week of gestation and matures through a complex process of neuronal migration and development. (1) By week 20, the corpus callosum can be seen using ultrasound examination or fetal MRI. (2) Although the corpus callosum may be considered fully developed at 4 years of age, as most neural structures, it likely continues to change in the following years of life. (3)

The corpus callosum has traditionally been viewed as having five distinct parts: the rostrum, genu, anterior midbody, isthmus, and splenium.The primary role of the corpus callosum is to facilitate communication between the hemispheres, using both inhibitory and excitory interhemispheric transfer. (4)

Colpocephaly represents a disproportionate enlargement of the occipital horns of the lateral ventricles. (5)This does not represent hydrocephalus but may represent the reduction of ipsilateral corti-

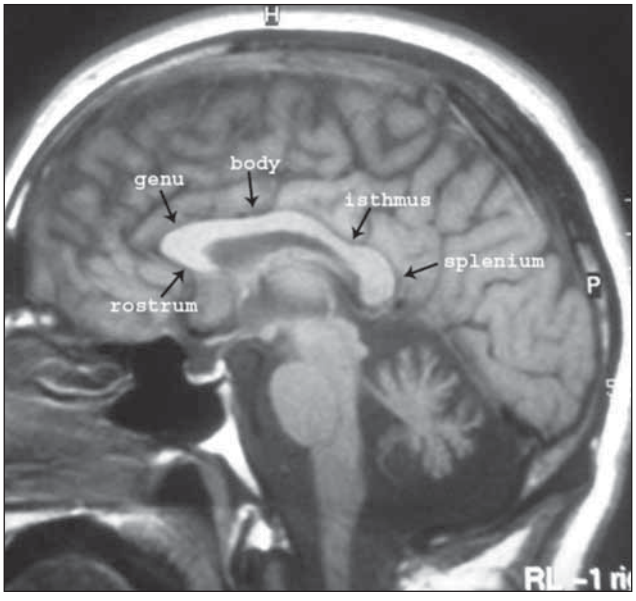

FIGURE 1. *Regions of intact corpus callosum. Images from: http://emedicine.medscape.com/article/407730-overview*

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cal association tracts. It is a nonspecific finding and it can be associated with agenesis of the corpus callosum, Chiari malformation, lissencephaly and microcephaly. The cause is maybe a disorder in neuronal migration during early brain development, perinatal injuries or other central nervous system disorders.(6)Clinicaly the patiens may present visual disturbances, spasticity and moderate mental retardation.

Corpus callosum agenesis (CCA) is among the most common brain malformations observed in humans.(2),(7). Its incidence varies as a function of both diagnostic techniques and sample populations: in the general population, its estimated prevalence is 3–7 per 1000 birth, while in children with developmental disabilities it is 2-3 per 100. (8)

It can occur either as an isolated malformation (isolated CCA) (49% of cases), or in association with another malformation syndrome, including malformation of the forebrain, secondary to intrauterine viral infections, toxic or metabolic disease, chromosomal abnormalities .(9)

CASE REPORT

T.F. is a 28 year-old woman, with no malformation diagnosed before birth (maybe no performant ultrasound was performed), with a normal birth but who was diagnosed at the age of 1 year and nine months with meningococcal meningitis. No imagistical investigations were performed at that time. She recovered, but continued with a history of frequent and severe headaches for which she was imagistically investigated at the age of 14 (in the year 2000).

The patient underwent a cerebral contrast enhanced MRI examination that showed a simetrical ventricular system, small anterior horns of the lateral ventricles, **dilatation of the posterior horns of the lateral ventricles, absence of the interventricular septum,** normal aqueduct and IV ventricle, almost entirely absence of corpus callosum except a small anterior fragment visible in 2 sagittal incidences.

The conclusion issued was dysgenesis of the corpus callosum and coroborated with the CT scan colpocephaly (dilatation of the posterior horns of the lateral ventricles).

The pacient lives in a city, studied psychology and is working in an office.

In april 2013 she was admitted in our clinic complaining of frontal and retro- orbital headaches, vertigo and head ”heaviness”. The headache is spontaneous, it isn’t associated with nausea, vomiting,

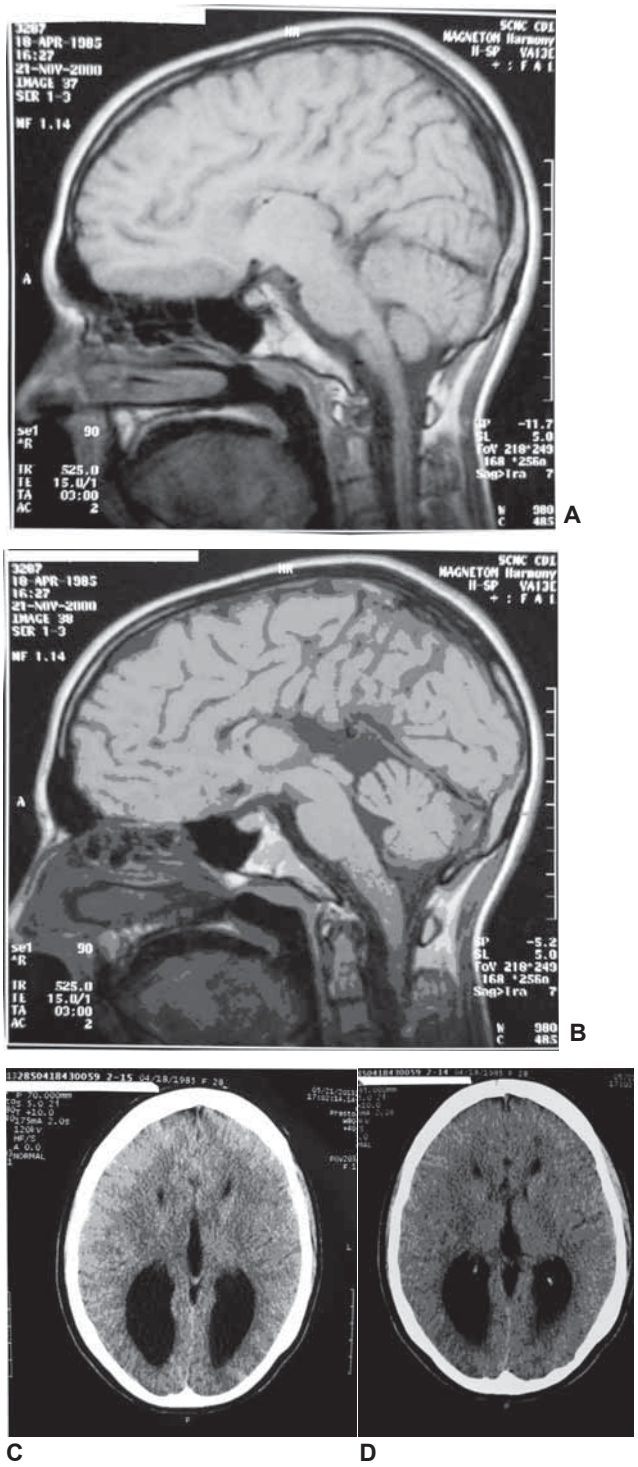

FIGURE 2. A. *Cerebral MRI (2000) – Sagital section showing almost entirely absence of corpus callosum except a small anterior fragment* ***B.*** *Cerebral MRI (2000)* ***C.*** *and* ***D.*** *Cerebral CT scan (2000) showing colpocephaly (dilatation of the posterior horns of the lateral ventricles).*

diurnal variation, fever, photophobia or phonophobia, and is relieved with usual analgesics.

On careful anamnesis we found out that the patient had suffered from depression and irritability, diminished interest in pleasurable activities, fatigue and loss of energy treated with antidepressives and

anxyolitics. There was a positive family history for migraine but none for any major medical or psychiatric disorders.

The neurologic examination was almost normal, except horizontal gaze nystagmus (right and left beating nystagmus) and abnormally increased osteotendinous reflexes - right bicipital, tricipital and styloradial and left rotulian.

A native cerebral CT scan performed in 2013 showed no recent heterodense lesions or an intracranial expansive process, slightly asymmetrical ventricular system with the dilatation of the posterior horns of lateral ventricles – colpocephaly.

The CT scan on the images taken revealed the absence of the corpus callosum (agenesia).

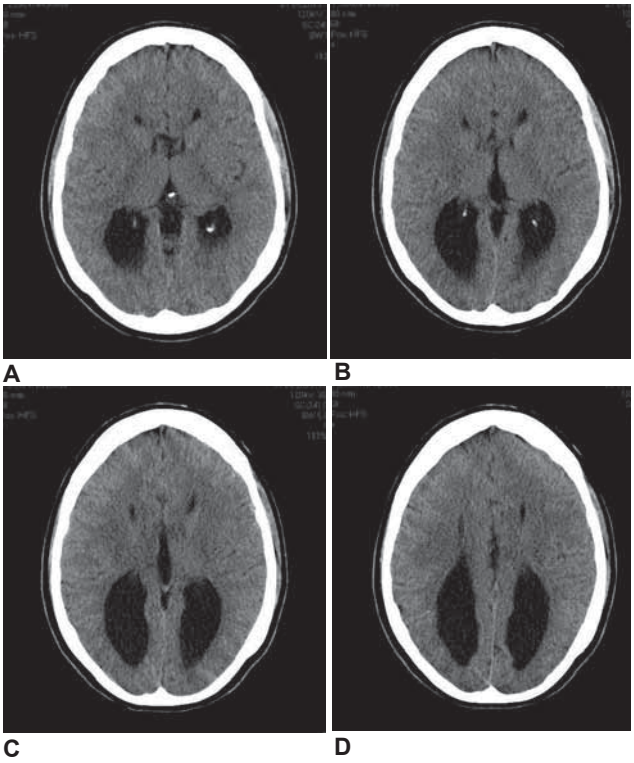

FIGURE 3. **A, B, C, D** *Cerebral CT scan (2013) – showing the absence of the corpus callosum (agenesia) and dilatation of the posterior horns of lateral ventricles* **–** *colpocephaly*

DISCUSSION

Neuroradiological assessment (CT or MRI) may reveal abnormalities that clinical examination do not even imagine. Patients with corpus calosum agenesis or dysgenesis may be asymptomatic and the malformation may be only discovered incidentally. The same for colpocephaly were MRI and CT scan reveals a dilation of the occipital horns and an abnormal separation of the frontal horns that are parallel; On the CT scan performed in 2013 the corpus callosum is not seen and the cortical gyri have a characteristic radiating disposition.

There are no specific treatments for agenesis or dysgenesis of the corpus callosum. In the case that there are clinical signs and symptoms, a complex team may include neurologist, neuropsychologist, occupational therapist, physiotherapist, speech and language therapist, paediatrician, geneticist, but also specific schooling support and early intervention specialists in order to help the patient.

CONCLUSIONS AND CASE PARTICULARITY

The diagnosis of corpus callosum dysgenesis as well as colpocephaly is given by neuroimaging. Magnetic resonance imaging (MRI) is currently the imaging procedure of choice, as the CT scan could not show the small part of the corpus calosum still present.

The patient we described had minimal stigmata of this disease, having no history of seizures, a good IQ, no spasticity or visual disturbancies. The meningococic meningitis is maybe responsible of her chronical headaches, but all the malformations were descovered accidentaly. We also inspected her mother's cerebral MRI and no malformations were described.

Prognosis depends on the extent and severity of malformations. It is not possible for the corpus callosum to regenerate or degenerate.

The patient will be imagistically monitored every 6-12 months, in order to identify any sign of activity of the colpocephaly. She was treated symptomaticaly.

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